

CLAIMS:

1. [Once Amended] An isolated, or recombinant or synthetic DNA molecule encoding
(a) a proteoglycan expressed in retinal tissue having a membrane anchor sequence and a leucine-rich repeat motif comprising the amino acid sequence: x-x-(I, V or L)-x-x-x-x-(F, P or L)-x-x-(L or P)-x-x-L-x-x-(L or I)-x-L-x-x-N-x-(I or L) where "x" represents any amino acid, wherein an impairment in function of the proteoglycan is associated with a complete CSNB phenotype;
(b) the amino acid sequence of SEQ ID NO: 2;
(b) the amino acid sequence of SEQ ID NO: 2 with conservative amino acid substitutions; or
(c) an amino acid sequence which is at least 50% homologous to SEQ ID NO: 2.

2. [Once Amended] The DNA molecule of claim 1 which is expressed in tissues including the kidney and the retina encodes an amino acid sequence which is at least 70% homologous to SEQ ID NO: 2.

3. The DNA molecule of claim 1 wherein said DNA is cDNA.

4. [deleted] The DNA molecule of claim 1 wherein said DNA is human DNA.

5. [deleted] The DNA molecule of claim 1 wherein said DNA is murine DNA.

6. [deleted] The DNA molecule of claim 1 wherein said DNA encodes nyctalopin; an amino acid sequence which is at least 50% homologous to nyctalopin; the amino acid sequence of SEQ ID NO: 2; or an amino acid sequence which is at least 50% homologous to SEQ ID NO: 2.

7. [deleted] The DNA molecule of claim 6 wherein said DNA encodes the amino acid sequence SEQ ID NO: 2 with conservative amino acid substitutions.

8. [Once Amended] The DNA molecule of claim 1 wherein said DNA has the nucleotide sequence corresponding to SEQ ID NO:1 or naturally occurring allelic variants of SEQ ID NO:1 nucleotide sequences corresponding to naturally occurring alleles thereof.

9. [Once Amended] An isolated or, recombinant or synthetic DNA molecule or polynucleotide comprising a nucleotide sequence corresponding to SEQ ID NO: 1, substantially homologous to SEQ ID NO:1 or a nucleotide sequence that hybridizes under highly stringent conditions to a hybridization probe having a nucleotide sequence of SEQ ID NO:1 or the complement of SEQ ID NO:1.
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10. The polynucleotide of claim 9 wherein said polynucleotide is selected from the group comprising:
(a) RNA;
(b) cDNA;
(c) genomic DNA; and
(d) synthetic nucleic acids.

11. [Once Amended] An expression vector comprising one of the DNAs or polynucleotides of claims 1-10, 2, 3, 8, 9 or 10.

12. A cultured cell comprising the expression vector of claim 11.

13. [Once Amended] A cultured cell comprising a the DNA sequence or polynucleotide of one of claims 1 to 10, operably linked to an expression control sequence. ✓

14. A cultured cell transfected with the vector of claim 11, or a progeny of said cell, wherein the cell expresses a mammalian GPI-anchored small leucine-rich proteoglycan.